



## LEPR gene

leptin receptor

### Normal Function

The *LEPR* gene provides instructions for making a protein called the leptin receptor, which is involved in the regulation of body weight. The leptin receptor protein is found on the surface of cells in many organs and tissues of the body, including a part of the brain called the hypothalamus. The hypothalamus controls hunger and thirst as well as other functions such as sleep, moods, and body temperature. It also regulates the release of many hormones that have functions throughout the body.

The leptin receptor is turned on (activated) by a hormone called leptin that attaches (binds) to the receptor, fitting into it like a key into a lock. Normally, the body's fat cells release leptin in proportion to their size. As fat cells become larger, they produce more leptin. This rise in leptin indicates that fat stores are increasing. In the hypothalamus, the binding of leptin to its receptor triggers a series of chemical signals that affect hunger and help produce a feeling of fullness (satiety).

### Health Conditions Related to Genetic Changes

#### leptin receptor deficiency

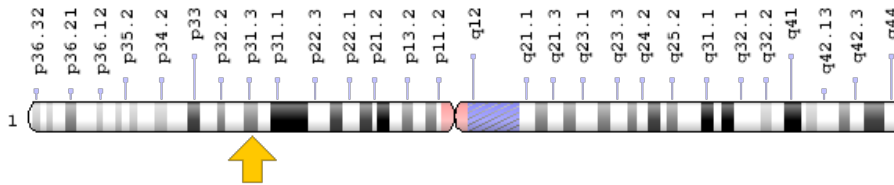
At least 18 *LEPR* gene mutations that cause leptin receptor deficiency have been identified; this disorder is associated with excessive hunger, massive weight gain, and reduced production of hormones that direct sexual development (hypogonadotropic hypogonadism). Some of the mutations result in less receptor protein getting to the cell surface where leptin binding takes place. The receptors that get to the cell surface may bind to leptin, but their signaling function is impaired. The resulting shortage of leptin signaling disrupts normal feelings of hunger and satiety, leading to extreme weight gain.

Because hypogonadotropic hypogonadism occurs in leptin receptor deficiency, researchers suggest that leptin receptor signaling is also involved in regulating the body's response to hormones that control sexual development, and that this response is affected by *LEPR* gene mutations. However, the mechanism of this effect is unknown.

## Chromosomal Location

Cytogenetic Location: 1p31.3, which is the short (p) arm of chromosome 1 at position 31.3

Molecular Location: base pairs 65,420,652 to 65,637,493 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- LEP-R
- LEPR\_HUMAN
- OB-R
- OB receptor
- OBR

## Additional Information & Resources

### Educational Resources

- Basic Neurochemistry: Molecular, Cellular and Medical Aspects (sixth edition, 1999): Neuronal Control of Food Intake  
<https://www.ncbi.nlm.nih.gov/books/NBK27993/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28LEPR%5BTI%5D%29+OR+%28leptin+receptor%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

### OMIM

- LEPTIN RECEPTOR  
<http://omim.org/entry/601007>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_LEPR.html](http://atlasgeneticsoncology.org/Genes/GC_LEPR.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=LEPR%5Bgene%5D>
- HGNC Gene Family: CD molecules  
<http://www.genenames.org/cgi-bin/genefamilies/set/471>
- HGNC Gene Family: Immunoglobulin like domain containing  
<http://www.genenames.org/cgi-bin/genefamilies/set/594>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=6554](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6554)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/3953>
- UniProt  
<http://www.uniprot.org/uniprot/P48357>

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<https://ghr.nlm.nih.gov/gene/LEPR>

Reviewed: July 2016

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services